



## GDAP1 gene

ganglioside induced differentiation associated protein 1

### Normal Function

The *GDAP1* gene provides instructions for making a protein called ganglioside-induced differentiation-associated protein 1. This protein is found in the outer membrane of mitochondria, the energy-producing centers within cells. Mitochondria are dynamic structures that change shape through processes called fission (splitting into smaller pieces) and fusion (combining pieces). Changes in shape are thought to be critical for mitochondria to work properly. Although the function of the *GDAP1* protein is not well understood, it appears to play a role in controlling the shape of mitochondria by promoting fission.

### Health Conditions Related to Genetic Changes

#### Charcot-Marie-Tooth disease

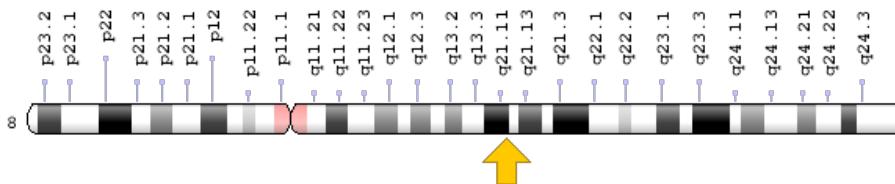
Researchers have identified more than 50 mutations in the *GDAP1* gene that can cause Charcot-Marie-Tooth disease, a disorder that results in muscle weakness, wasting (atrophy) of muscles, and loss of sensation in the feet, legs, and hands. Some of these mutations create a premature stop signal in the instructions for making the *GDAP1* protein. As a result, cells produce an abnormally short protein that is unlikely to be inserted correctly into the outer membrane of mitochondria. Other mutations change single building blocks (amino acids) used to make the *GDAP1* protein, which may alter its shape or function.

It is unclear how *GDAP1* gene mutations lead to the characteristic features of Charcot-Marie-Tooth disease. These mutations probably impair the mitochondria's ability to change shape, but how this impairment affects the peripheral nerves is not well understood.

## Chromosomal Location

Cytogenetic Location: 8q21.11, which is the long (q) arm of chromosome 8 at position 21.11

Molecular Location: base pairs 74,349,251 to 74,488,867 on chromosome 8 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- CMT2G
- CMT2K
- CMT4
- CMT4A
- ganglioside-induced differentiation-associated protein 1
- GDAP1\_HUMAN

## Additional Information & Resources

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Dynamic mitochondrial reticulum  
<https://www.ncbi.nlm.nih.gov/books/NBK26924/figure/A2605/>

### GeneReviews

- Charcot-Marie-Tooth Neuropathy Type 2  
<https://www.ncbi.nlm.nih.gov/books/NBK1285>
- Charcot-Marie-Tooth Neuropathy Type 4A  
<https://www.ncbi.nlm.nih.gov/books/NBK1539>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28GDAP1%5BTIAB%5D%29+OR+%28ganglioside-induced+differentiation-associated+protein+1%5BTIAB%5D%29%29+OR+%28CMT2G%5BTIAB%5D%29+OR+%28CMT2K%5BTIAB%5D%29+OR+%28CMT4A%5BTIAB%5D%29%29+AND+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- GANGLIOSIDE-INDUCED DIFFERENTIATION-ASSOCIATED PROTEIN 1  
<http://omim.org/entry/606598>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_GDAP1.html](http://atlasgeneticsoncology.org/Genes/GC_GDAP1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=GDAP1%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=15968](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=15968)
- Inherited Peripheral Neuropathies Mutation Database  
<http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=20>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/54332>
- UniProt  
<http://www.uniprot.org/uniprot/Q8TB36>

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